



IFWO

RAW SEQUENCE LISTING

DATE: 09/17/2004

PATENT APPLICATION: US/10/766,760

TIME: 15:44:58

Input Set : A:\Sequence Listing.TXT

Output Set: N:\CRF4\09172004\J766760.raw

4 <110> APPLICANT: Klein, Michael
5 Schultz, Dennis
7 <120> TITLE OF INVENTION: Gene Mutation Associated with
8 Age-Related Macular Degeneration
11 <130> FILE REFERENCE: 49321-117
13 <140> CURRENT APPLICATION NUMBER: 10/766,760
C--> 14 <141> CURRENT FILING DATE: 2004-01-27
16 <150> PRIOR APPLICATION NUMBER: 60/443,214
17 <151> PRIOR FILING DATE: 2003-01-27
19 <160> NUMBER OF SEQ ID NOS: 186
21 <170> SOFTWARE: FastSEQ for Windows Version 4.0
23 <210> SEQ ID NO: 1
24 <211> LENGTH: 18206
25 <212> TYPE: DNA
26 <213> ORGANISM: Homo sapiens
28 <220> FEATURE:
29 <221> NAME/KEY: CDS
30 <222> LOCATION: (230)...(17137)
32 <221> NAME/KEY: variation
33 <222> LOCATION: (14197)...(14197)
34 <223> OTHER INFORMATION: A and G allelic variation exists at this position
W--> 36 <221> variation
37 <222> LOCATION: (17811)...(17811)
38 <223> OTHER INFORMATION: C and T allelic variation exists at this position
W--> 40 <400> 1
41 gaagccgcat ccagacaaaa gctgccgcat cctgccctg cccaacccct ggagggattc 60
42 gagtttggtg cttgtccccg tctgattctc agcgccaaac tttttgctag ttcagagatt 120
43 ccaagagtct gatgagttac tctgagagga aaccctctgc ctgttggtga ggaggactga 180
44 gcacagtgct taggcgctga gggggaaaaa gagggggaaa aaaaagaaa atg att tcc 238
45 Met Ile Ser
46 1
48 tgg gaa gtt gtc cat aca gta ttc ctg ttt gct ctt ctt tat tct tcc 286
49 Trp Glu Val Val His Thr Val Phe Leu Phe Ala Leu Leu Tyr Ser Ser
50 5 10 15
52 cta gct caa gat gcg agc ccc cag tca gag atc aga gct gag gaa att 334
53 Leu Ala Gln Asp Ala Ser Pro Gln Ser Glu Ile Arg Ala Glu Glu Ile
54 20 25 30 35
56 ccc gag ggg gcc tcc acg ttg gct ttt gtg ttt gat gtg act ggt tct 382
57 Pro Glu Gly Ala Ser Thr Leu Ala Phe Val Phe Asp Val Thr Gly Ser
58 40 45 50
60 atg tat gat gat tta gtt cag gtg att gaa ggg gct tcc aaa att ttg 430
61 Met Tyr Asp Asp Leu Val Gln Val Ile Glu Gly Ala Ser Lys Ile Leu
62 55 60 65

ENTERED

RAW SEQUENCE LISTING

DATE: 09/17/2004

PATENT APPLICATION: US/10/766,760

TIME: 15:44:58

Input Set : A:\Sequence Listing.TXT

Output Set: N:\CRF4\09172004\J766760.raw

```

64 gag acg tct ttg aaa aga cct aaa aga cct ctt ttc aac ttt gcg ttg 478
65 Glu Thr Ser Leu Lys Arg Pro Lys Arg Pro Leu Phe Asn Phe Ala Leu
66      70      75      80
68 gtg cct ttc cat gat cca gaa att ggc cca gtg aca att acc aca gat 526
69 Val Pro Phe His Asp Pro Glu Ile Gly Pro Val Thr Ile Thr Thr Asp
70      85      90      95
72 ccc aag aaa ttt caa tat gaa ctg aga gaa ctg tat gtt cag ggt ggt 574
73 Pro Lys Lys Phe Gln Tyr Glu Leu Arg Glu Leu Tyr Val Gln Gly Gly
74 100      105      110      115
76 ggt gat tgc cca gaa atg agt att gga gct ata aaa att gcc ttg gaa 622
77 Gly Asp Cys Pro Glu Met Ser Ile Gly Ala Ile Lys Ile Ala Leu Glu
78      120      125      130
80 att tct ctt cct ggt tct ttc atc tat gtt ttc act gat gct cgg tcc 670
81 Ile Ser Leu Pro Gly Ser Phe Ile Tyr Val Phe Thr Asp Ala Arg Ser
82      135      140      145
84 aaa gat tac cgg ctg acc cat gag gtg ctg caa ctt atc caa cag aaa 718
85 Lys Asp Tyr Arg Leu Thr His Glu Val Leu Gln Leu Ile Gln Gln Lys
86      150      155      160
88 cag tca caa gtc gta ttt gtt ctg act gga gat tgt gat gac agg acc 766
89 Gln Ser Gln Val Val Phe Val Leu Thr Gly Asp Cys Asp Asp Arg Thr
90      165      170      175
92 cat att gga tat aaa gtc tat gaa gaa att gcc tct aca agt tct ggt 814
93 His Ile Gly Tyr Lys Val Tyr Glu Glu Ile Ala Ser Thr Ser Ser Gly
94 180      185      190      195
96 caa gtg ttc cat ctg gac aaa aaa caa gtt aat gag gta tta aaa tgg 862
97 Gln Val Phe His Leu Asp Lys Lys Gln Val Asn Glu Val Leu Lys Trp
98      200      205      210
100 gta gaa gaa gca gta cag gcc tcc aaa gtt cac ctt tta tcc aca gat 910
101 Val Glu Glu Ala Val Gln Ala Ser Lys Val His Leu Leu Ser Thr Asp
102      215      220      225
104 cat ttg gaa cag gct gta aat act tgg aga att cct ttt gat ccc agc 958
105 His Leu Glu Gln Ala Val Asn Thr Trp Arg Ile Pro Phe Asp Pro Ser
106      230      235      240
108 ctg aaa gag gtc act gtg tct ttg agt ggg cct tct cca atg att gaa 1006
109 Leu Lys Glu Val Thr Val Ser Leu Ser Gly Pro Ser Pro Met Ile Glu
110      245      250      255
112 att cgc aat cct tta ggg aag ctg ata aaa aag gga ttt ggc ctg cat 1054
113 Ile Arg Asn Pro Leu Gly Lys Leu Ile Lys Lys Gly Phe Gly Leu His
114 260      265      270      275
116 gag cta tta aat atc cat aac tct gcc aaa gta gtg aat gtg aaa gag 1102
117 Glu Leu Leu Asn Ile His Asn Ser Ala Lys Val Val Asn Val Lys Glu
118      280      285      290
120 cca gag gct gga atg tgg aca gtg aag acc tca agc agt gga agg cac 1150
121 Pro Glu Ala Gly Met Trp Thr Val Lys Thr Ser Ser Ser Gly Arg His
122      295      300      305
124 tct gtt cgc att act ggc ctg agt act att gat ttc cga gct ggc ttt 1198
125 Ser Val Arg Ile Thr Gly Leu Ser Thr Ile Asp Phe Arg Ala Gly Phe
126      310      315      320
128 tct cga aag ccc acc ctg gac ttc aaa aaa aca gtc agc aga cca gtg 1246

```

RAW SEQUENCE LISTING

PATENT APPLICATION: US/10/766,760

DATE: 09/17/2004

TIME: 15:44:58

Input Set : A:\Sequence Listing.TXT

Output Set: N:\CRF4\09172004\J766760.raw

```

129 Ser Arg Lys Pro Thr Leu Asp Phe Lys Lys Thr Val Ser Arg Pro Val
130      325      330      335
132 caa gga ata cct acc tat gta ctg ctc aat act tct gga att tcc act 1294
133 Gln Gly Ile Pro Thr Tyr Val Leu Leu Asn Thr Ser Gly Ile Ser Thr
134 340      345      350      355
136 cca gct aga ata gat ctt ctt gaa ctt ttg agt atc tca gga agt tct 1342
137 Pro Ala Arg Ile Asp Leu Leu Glu Leu Leu Ser Ile Ser Gly Ser Ser
138      360      365      370
140 ctt aag act att cct gtt aaa tat tac cca cat cga aaa cct tat ggc 1390
141 Leu Lys Thr Ile Pro Val Lys Tyr Tyr Pro His Arg Lys Pro Tyr Gly
142      375      380      385
144 ata tgg aat att tct gac ttt gta cca cca aat gaa gct ttc ttt ctc 1438
145 Ile Trp Asn Ile Ser Asp Phe Val Pro Pro Asn Glu Ala Phe Phe Leu
146      390      395      400
148 aaa gta aca ggc tat gat aaa gat gat tac ctc ttc cag aga gta tca 1486
149 Lys Val Thr Gly Tyr Asp Lys Asp Asp Tyr Leu Phe Gln Arg Val Ser
150      405      410      415
152 agt gtt tcc ttt tct agt att gtc cca gat gct ccc aaa gtt acg atg 1534
153 Ser Val Ser Phe Ser Ser Ile Val Pro Asp Ala Pro Lys Val Thr Met
154 420      425      430      435
156 cct gag aaa acc cca gga tac tat ctg cag ccg ggc caa att ccc tgc 1582
157 Pro Glu Lys Thr Pro Gly Tyr Tyr Leu Gln Pro Gly Gln Ile Pro Cys
158      440      445      450
160 tct gtt gac agt ctt ttg ccc ttt acc ttg agc ttt gtc aga aat gga 1630
161 Ser Val Asp Ser Leu Leu Pro Phe Thr Leu Ser Phe Val Arg Asn Gly
162      455      460      465
164 gtt aca ctt gga gta gac cag tat ttg aaa gaa tct gcc agt gtg aac 1678
165 Val Thr Leu Gly Val Asp Gln Tyr Leu Lys Glu Ser Ala Ser Val Asn
166      470      475      480
168 tta gat att gca aag gtc act ttg tct gac gaa ggt ttc tat gaa tgc 1726
169 Leu Asp Ile Ala Lys Val Thr Leu Ser Asp Glu Gly Phe Tyr Glu Cys
170      485      490      495
172 att gct gtc agc agt gca ggt act gga cgg gca cag aca ttt ttt gac 1774
173 Ile Ala Val Ser Ser Ala Gly Thr Gly Arg Ala Gln Thr Phe Phe Asp
174 500      505      510      515
176 gta tca gag ccc cct ccg gtc atc caa gtg cct aac aat gtt aca gtc 1822
177 Val Ser Glu Pro Pro Pro Val Ile Gln Val Pro Asn Asn Val Thr Val
178      520      525      530
180 act cct gga gag aga gca gtt tta aca tgt ctc atc atc agt gcg gtg 1870
181 Thr Pro Gly Glu Arg Ala Val Leu Thr Cys Leu Ile Ile Ser Ala Val
182      535      540      545
184 gat tac aat cta acc tgg cag agg aat gac aga gat gtc aga ctg gca 1918
185 Asp Tyr Asn Leu Thr Trp Gln Arg Asn Asp Arg Asp Val Arg Leu Ala
186      550      555      560
188 gag cca gcg aga att agg acc ttg gct aat ctg tca ttg gag cta aag 1966
189 Glu Pro Ala Arg Ile Arg Thr Leu Ala Asn Leu Ser Leu Glu Leu Lys
190      565      570      575
192 agt gtg aaa ttc aac gat gct gga gag tat cat tgt atg gtt tct agt 2014
193 Ser Val Lys Phe Asn Asp Ala Gly Glu Tyr His Cys Met Val Ser Ser

```

RAW SEQUENCE LISTING

DATE: 09/17/2004

PATENT APPLICATION: US/10/766,760

TIME: 15:44:58

Input Set : A:\Sequence Listing.TXT

Output Set: N:\CRF4\09172004\J766760.raw

194	580		585		590		595	
196	gaa ggt gga tca tca gcc gct tca gtt ttc ctc aca gtg caa gaa cca	2062						
197	Glu Gly Gly Ser Ser Ala Ala Ser Val Phe Leu Thr Val Gln Glu Pro							
198			600		605		610	
200	ccc aaa gtc act gtg atg ccc aag aat cag tct ttc aca gga ggg tct	2110						
201	Pro Lys Val Thr Val Met Pro Lys Asn Gln Ser Phe Thr Gly Gly Ser							
202			615		620		625	
204	gag gtc tcc atc atg tgt tct gca aca ggt tat ccc aaa cca aag att	2158						
205	Glu Val Ser Ile Met Cys Ser Ala Thr Gly Tyr Pro Lys Pro Lys Ile							
206			630		635		640	
208	gcc tgg acc gtt aac gat atg ttt atc gtg ggt tca cac agg tat agg	2206						
209	Ala Trp Thr Val Asn Asp Met Phe Ile Val Gly Ser His Arg Tyr Arg							
210			645		650		655	
212	atg acc tca gat ggt acc tta ttt atc aaa aat gca gct ccc aaa gat	2254						
213	Met Thr Ser Asp Gly Thr Leu Phe Ile Lys Asn Ala Ala Pro Lys Asp							
214	660		665		670		675	
216	gca ggg atc tat ggt tgc cta gca agt aat tca gct gga aca gat aaa	2302						
217	Ala Gly Ile Tyr Gly Cys Leu Ala Ser Asn Ser Ala Gly Thr Asp Lys							
218			680		685		690	
220	cag aat tct act ctc aga tac att gaa gcc cct aag ttg atg gta gtt	2350						
221	Gln Asn Ser Thr Leu Arg Tyr Ile Glu Ala Pro Lys Leu Met Val Val							
222			695		700		705	
224	cag agt gag ctc ttg gtt gcc ctt ggg gat ata acc gtt atg gaa tgc	2398						
225	Gln Ser Glu Leu Leu Val Ala Leu Gly Asp Ile Thr Val Met Glu Cys							
226			710		715		720	
228	aaa acc tct ggt att cct cca cct caa gtt aaa tgg ttc aaa gga gat	2446						
229	Lys Thr Ser Gly Ile Pro Pro Pro Gln Val Lys Trp Phe Lys Gly Asp							
230			725		730		735	
232	ctt gag ttg agg ccc tca aca ttc ctc att att gac cct ctc ttg gga	2494						
233	Leu Glu Leu Arg Pro Ser Thr Phe Leu Ile Ile Asp Pro Leu Leu Gly							
234	740		745		750		755	
236	ctt ttg aag att caa gaa aca caa gat ctg gat gct ggc gat tat acc	2542						
237	Leu Leu Lys Ile Gln Glu Thr Gln Asp Leu Asp Ala Gly Asp Tyr Thr							
238			760		765		770	
240	tgt gta gcc atc aat gag gct gga aga gca act ggc aag ata act ctg	2590						
241	Cys Val Ala Ile Asn Glu Ala Gly Arg Ala Thr Gly Lys Ile Thr Leu							
242			775		780		785	
244	gat gtt ggc tca cct cca gtt ttc ata caa gaa cct gct gat gtg tct	2638						
245	Asp Val Gly Ser Pro Pro Val Phe Ile Gln Glu Pro Ala Asp Val Ser							
246			790		795		800	
248	atg gaa att ggc tca aat gtg aca tta cct tgt tat gtt cag ggt tat	2686						
249	Met Glu Ile Gly Ser Asn Val Thr Leu Pro Cys Tyr Val Gln Gly Tyr							
250			805		810		815	
252	cca gaa cca aca atc aaa tgg cga aga tta gac aac atg cca att ttc	2734						
253	Pro Glu Pro Thr Ile Lys Trp Arg Arg Leu Asp Asn Met Pro Ile Phe							
254	820		825		830		835	
256	tca aga cct ttt tca gtt agt tcc atc agc caa cta aga aca gga gct	2782						
257	Ser Arg Pro Phe Ser Val Ser Ser Ile Ser Gln Leu Arg Thr Gly Ala							
258			840		845		850	

RAW SEQUENCE LISTING

DATE: 09/17/2004

PATENT APPLICATION: US/10/766,760

TIME: 15:44:58

Input Set : A:\Sequence Listing.TXT

Output Set: N:\CRF4\09172004\J766760.raw

```

260 ctc ttt att tta aac tta tgg gca agt gat aaa gga acc tat att tgt 2830
261 Leu Phe Ile Leu Asn Leu Trp Ala Ser Asp Lys Gly Thr Tyr Ile Cys
262      855      860      865
264 gaa gct gaa aac cag ttt gga aag atc cag tca gag aca aca gta aca 2878
265 Glu Ala Glu Asn Gln Phe Gly Lys Ile Gln Ser Glu Thr Thr Val Thr
266      870      875      880
268 gtg acc gga ctt gtt gct cca ctt att gga atc agc cct tca gtg gcc 2926
269 Val Thr Gly Leu Val Ala Pro Leu Ile Gly Ile Ser Pro Ser Val Ala
270      885      890      895
272 aat gtt att gaa gga cag cag ctt act ttg ccc tgt act ctg tta gct 2974
273 Asn Val Ile Glu Gly Gln Gln Leu Thr Leu Pro Cys Thr Leu Leu Ala
274 900      905      910      915
276 gga aat ccc att cca gaa cgt cgg tgg att aag aat tca gct atg ttg 3022
277 Gly Asn Pro Ile Pro Glu Arg Arg Trp Ile Lys Asn Ser Ala Met Leu
278      920      925      930
280 ctc caa aat cct tac atc act gtg cgc agt gat ggg agc ctc cat att 3070
281 Leu Gln Asn Pro Tyr Ile Thr Val Arg Ser Asp Gly Ser Leu His Ile
282      935      940      945
284 gaa aga gtt cag ctt cag gat ggt ggt gaa tat act tgt gtg gcc agt 3118
285 Glu Arg Val Gln Leu Gln Asp Gly Gly Glu Tyr Thr Cys Val Ala Ser
286      950      955      960
288 aac gtt gct ggg acc aat aac aaa act acc tct gtg gtt gtg cat gtt 3166
289 Asn Val Ala Gly Thr Asn Asn Lys Thr Thr Ser Val Val Val His Val
290      965      970      975
292 ctg cca acc att cag cat ggg cag cag ata ctc agt aca att gaa ggc 3214
293 Leu Pro Thr Ile Gln His Gly Gln Gln Ile Leu Ser Thr Ile Glu Gly
294 980      985      990      995
296 att cca gta act tta cca tgc aaa gca agt gga aat ccc aaa ccg tct 3262
297 Ile Pro Val Thr Leu Pro Cys Lys Ala Ser Gly Asn Pro Lys Pro Ser
298      1000      1005      1010
300 gtc atc tgg tcc aag aaa gga gag ctg att tca acc agc agt gct aag 3310
301 Val Ile Trp Ser Lys Lys Gly Glu Leu Ile Ser Thr Ser Ser Ala Lys
302      1015      1020      1025
304 ttt tca gca gga gct gat ggt agt ctg tat gtg gta tca cct gga gga 3358
305 Phe Ser Ala Gly Ala Asp Gly Ser Leu Tyr Val Val Ser Pro Gly Gly
306      1030      1035      1040
308 gag gag agt ggg gag tat gtc tgc act gcc acc aat aca gcc ggc tac 3406
309 Glu Glu Ser Gly Glu Tyr Val Cys Thr Ala Thr Asn Thr Ala Gly Tyr
310      1045      1050      1055
312 gcc aaa agg aaa gtg cag cta aca gtc tat gta agg ccc aga gtg ttt 3454
313 Ala Lys Arg Lys Val Gln Leu Thr Val Tyr Val Arg Pro Arg Val Phe
314 1060      1065      1070      1075
316 gga gat caa cga gga ctg tcc cag gat aag cct gtt gag atc tcc gtc 3502
317 Gly Asp Gln Arg Gly Leu Ser Gln Asp Lys Pro Val Glu Ile Ser Val
318      1080      1085      1090
320 ctt gca ggg gaa gag gta aca ctt cca tgt gaa gtg aag agc tta cct 3550
321 Leu Ala Gly Glu Glu Val Thr Leu Pro Cys Glu Val Lys Ser Leu Pro
322      1095      1100      1105
324 cca ccc ata att act tgg gcc aaa gaa acc cag ctc atc tca ccg ttc 3598

```

VERIFICATION SUMMARY

PATENT APPLICATION: US/10/766,760

DATE: 09/17/2004

TIME: 15:44:59

Input Set : A:\Sequence Listing.TXT

Output Set: N:\CRF4\09172004\J766760.raw

L:14 M:271 C: Current Filing Date differs, Replaced Current Filing Date
L:36 M:258 W: Mandatory Feature missing, <220> Tag not found for SEQ ID#:1
L:40 M:258 W: Mandatory Feature missing, <220> Tag not found for SEQ ID#:1
L:1460 M:336 W: Invalid Amino Acid Number in Coding Region, SEQ ID:1